

APPENDIX I
CURRENTLY PENDING CLAIMS

WHAT IS CLAIMED IS:

- 1 1. A method for comparing a test genome to a reference genome, said
2 method comprising:
 - 3 (i) providing a plurality of clones of known size that substantially cover at
4 least a portion of said test genome;
 - 5 (ii) obtaining sequence information from the termini of each of said plurality
6 of clones, thereby obtaining a pair of terminal sequences;
 - 7 (iii) identifying a pair of sequences within said reference genome that
8 corresponds to each of said pairs of terminal sequences; and
 - 9 (iv) determining the relationship between the members of each pair of
10 corresponding sequences within said reference genome;
11 wherein a difference in the observed relationship between the members of any
12 of said pairs of corresponding sequences within said reference genome and the expected
13 relationship based upon said known size of said plurality of clones indicates the presence of a
14 rearrangement in said test genome compared to said reference genome.
- 1 2. The method of claim 1, further comprising determining the sequence
2 of said test genome over a region spanning at least one breakpoint of said rearrangement.
- 1 3. The method of claim 1, wherein said reference genome is a human
2 genome.
- 1 4. The method of claim 1, wherein said test genome is from a tumor cell.
- 1 5. The method of claim 1, wherein said reference genome and said test
2 genome are from different species.
- 1 6. The method of claim 1, wherein said plurality of clones covers
2 substantially all of said test genome.

1 7. The method of claim 1, wherein the members of at least one pair of
2 corresponding sequences within said reference genome are closer together than expected
3 based on said known size of said plurality of clones, indicating the presence of an insertion in
4 said test genome between the pair of terminal sequences corresponding to said at least one
5 pair of corresponding sequences.

1 8. The method of claim 1, wherein the members of at least one pair of
2 corresponding sequences within said reference genome are further apart than expected based
3 on said known size of said plurality of clones, indicating the presence of a deletion in said
4 test genome between the pair of terminal sequences corresponding to said at least one pair of
5 corresponding sequences.

1 9. The method of claim 1, wherein the members of at least one pair of
2 corresponding sequences within said reference genome are present on different chromosomes
3 within said reference genome, indicating the presence of a translocation in said test genome
4 between the pair of terminal sequences corresponding to said at least one pair of
5 corresponding sequences.

1 10. The method of claim 1, further comprising determining the frequency
2 of each of said terminal sequences within said plurality of clones, wherein an increased or
3 decreased relative frequency of any of said terminal sequences indicates the presence of an
4 amplification or a deletion in said test genome that includes said terminal sequence.

1 19. The method of claim 1, wherein said reference genome is a human
2 genome, and wherein said plurality of clones comprises at least about 100,000 clones.

1 20. The method of claim 19, wherein said plurality of clones comprises at
2 least about 200,000 clones.

1 21. The method of claim 20, wherein said plurality of clones comprises at
2 least about 250,000 clones.

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1 22. The method of claim 1, wherein said terminal sequences are
2 determined by automated sequencing.

1 23. The method of claim 1, wherein said pairs of terminal sequences from
2 said test genome are compared to said pairs of corresponding sequences within said reference
3 genome using a computer.